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DUPLICATION Xq25-27.1 IN A GIRL WITH SHORT STATURE AND DYSMORPHIC FEATURES : EMERGING PHENOTYPES OF CRYPTIC DISTAL Xq DUPLICATIONS

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Large, cytogenetically visible duplications of the long arm of the X-chromosome are rare, but with the recent CGH/micro-array techniques more and smaller imbalances can be detected.

Prevalence of Xq duplications is unknown, but about 40 cases of cytogenetically visible cases have been reported and about 50 cases of cryptic duplications, mostly encompassing the MECP2 gene on Xq28, being an important dosage sensitive gene on Xq. Duplications of this region yield a recognisable phenotype including hypotonia, developmental delay, proneness to infections and distinctive facial features.

Clinical expression is gender dependent because of X-inactivation, with usually milder symptoms in females than in males. For several reasons there can be failure of X chromosome dosage compensation in females, giving rise to functional disomie and clinical symptoms.

The discovery of more cryptic duplications allows the description of some more specific, recognisable phenotypes.

We describe a female patient with a duplication of Xq25-27.1, encompassing the SOX3 gene. The girl showed severe growth retardation, feeding problems and dysmorphic features, like micrognathia, large, dysplastic ears and camptodactyly of 5th fingers. Overlapping with this deletion in our patient, several patients have been reported in the literature with some phenotypic similarities to our case.

We discuss the phenotype of similar duplications in boys, the role the SOX3 gene and the possible mechanisms leading to the phenotype in girls with Xq duplications.